Disease Name — Argininosuccinyl-CoA lyase deficiency

Alternate name(s)	Argininosuccinase deficiency, Argininosuccinic aciduria, Argininosuccinic acid lyase deficiency, ASL deficiency
Acronym	ASAL
Disease Classification	Amino Acid Disorder
Variants	Yes
Variant name	Late onset form
Symptom onset	Neonatal onset is typical, although later-onset may occur.
Symptoms	Anorexia, vomiting, lethargy, seizures and coma possibly leading to death.
Natural history without treatment	Mental and physical retardation due to hyperammonemia, cyclic vomiting, seizures, cerebral edema and trichorrhexis nodosa. Coma and death possible.
Natural history with treatment	Normal mental and physical development is possible if treatment is initiated before hyperammonemic crisis.
Treatment	Protein restricted diet, arginine supplementation to help complete the urea cycle, essential amino acid supplementation, ammonia scavenging drugs in some cases and supplemental carnitine if patient has a secondary deficiency.
Other	Enzyme is genetically heterogeneous and patients may present in infancy/childhood with MR or seizures.
Physical phenotype	Trichorrhexis nodosa (short, dry, brittle hair) in older patients.
Inheritance	Autosomal recessive
General population incidence	1:70,000
Ethnic differences	No
Population	N/A
Ethnic incidence	N/A
Enzyme location	Erythrocytes, liver and fibroblasts
Enzyme Function	Catalyzes the conversion of argininosuccinate to fumurate and arginine as part of the urea cycle.
Missing Enzyme	Argininosuccinate lyase
Metabolite changes	Hyperammonemia
Gene	ASL
Gene location	7q11.2
DNA testing available	No
DNA testing detail	No common mutation known. More that 25 mutations detected.
Prenatal testing	Enzyme assay in cultured amniocytes. DNA possible if mutations known. Analyte testing of amniocytes.
MS/MS Profile	Citrulline is elevated, may show elevated argininosuccinic peak.
OMIM Link	http://www.ncbi.nlm.nih.gov/entrez/dispomim.cgi?id=207900
Genetests Link	www.genetests.org
Support Group	National Urea Cycle Disorders Foundation http://www.nucdf.org/ National Coalition for PKU and Allied Disorders http://www.pku-allieddisorders.org/ Children Living with Inherited Metabolic Diseases http://www.climb.org.uk/